



YARS gene

tyrosyl-tRNA synthetase

Normal Function

The *YARS* gene provides instructions for making an enzyme called tyrosyl-tRNA synthetase. This enzyme is found in all cells, where it plays an important role in the production (synthesis) of proteins. During protein synthesis, building blocks (amino acids) are connected together in a specific order, creating the chain of amino acids that makes up the protein. Tyrosyl-tRNA synthetase plays a role in adding the amino acid tyrosine at the proper place in a protein's chain of amino acids.

In addition to its role in protein synthesis, tyrosyl-tRNA synthetase appears to have other functions. Under certain conditions, such as inflammation, this enzyme is cut (cleaved) into two fragments called mini-tyrRS and C-tyrRS. Research indicates that mini-tyrRS promotes the growth of new blood vessels (angiogenesis). Both fragments appear to stimulate the movement of particular cells, such as white blood cells that help fight infection.

Health Conditions Related to Genetic Changes

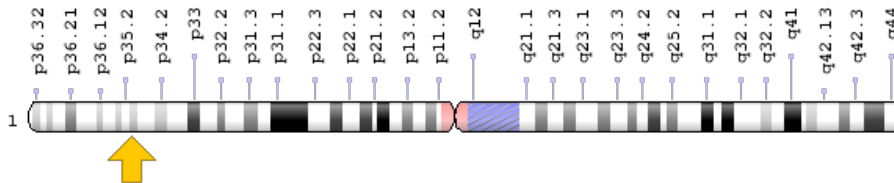
Charcot-Marie-Tooth disease

At least three mutations in the *YARS* gene cause a form of Charcot-Marie-Tooth disease known as dominant intermediate C. One mutation replaces the amino acid glycine with the amino acid arginine at protein position 41 of the tyrosyl-tRNA synthetase enzyme (written as Gly41Arg or G41R). Another mutation replaces the amino acid glutamic acid with the amino acid lysine at protein position 196 (written as Glu196Lys or E196K). A third *YARS* gene mutation results in an altered version of the tyrosyl-tRNA synthetase enzyme that is missing four amino acids. Mutations in the *YARS* gene probably reduce the activity of tyrosyl-tRNA synthetase, which could affect the synthesis of any protein that contains tyrosine. It is unclear how these mutations lead to the dominant intermediate C form of Charcot-Marie-Tooth disease.

Chromosomal Location

Cytogenetic Location: 1p35.1, which is the short (p) arm of chromosome 1 at position 35.1

Molecular Location: base pairs 32,775,237 to 32,818,032 on chromosome 1 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- SYYC_HUMAN
- Tyrosyl-tRNA Ligase
- tyrRS
- YRS
- YTS

Additional Information & Resources

Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): Specific Enzymes Couple Each Amino Acid to Its Appropriate tRNA Molecule
<https://www.ncbi.nlm.nih.gov/books/NBK26829/#A1062>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28YARS%5BTIAB%5D%29+OR+%28tyrosyl-tRNA+synthetase%5BALL%5D%29%29+OR+%28tyrosyl+tRNA+ligase%5BALL%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

OMIM

- TYROSYL-tRNA SYNTHETASE
<http://omim.org/entry/603623>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_YARS.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=YARS%5Bgene%5D>
- HGNC Gene Family: Aminoacyl tRNA synthetases, Class I
<http://www.genenames.org/cgi-bin/genefamilies/set/131>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=12840
- Inherited Peripheral Neuropathies Mutation Database
<http://www.molgen.ua.ac.be/CMTMutations/Mutations/Mutations.cfm?Context=40>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/8565>
- UniProt
<http://www.uniprot.org/uniprot/P54577>

Sources for This Summary

- Antonellis A, Green ED. The role of aminoacyl-tRNA synthetases in genetic diseases. *Annu Rev Genomics Hum Genet.* 2008;9:87-107. doi: 10.1146/annurev.genom.9.081307.164204. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/18767960>
- Ewalt KL, Schimmel P. Activation of angiogenic signaling pathways by two human tRNA synthetases. *Biochemistry.* 2002 Nov 12;41(45):13344-9. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/12416978>
- Jordanova A, Irobi J, Thomas FP, Van Dijck P, Meerschaert K, Dewil M, Dierick I, Jacobs A, De Vriendt E, Guergueltcheva V, Rao CV, Tournev I, Gondim FA, D'Hooghe M, Van Gerwen V, Callaerts P, Van Den Bosch L, Timmermans JP, Robberecht W, Gettemans J, Thevelein JM, De Jonghe P, Kremensky I, Timmerman V. Disrupted function and axonal distribution of mutant tyrosyl-tRNA synthetase in dominant intermediate Charcot-Marie-Tooth neuropathy. *Nat Genet.* 2006 Feb; 38(2):197-202. Epub 2006 Jan 22.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/16429158>

- Wakasugi K, Slike BM, Hood J, Otani A, Ewalt KL, Friedlander M, Cheres DA, Schimmel P. A human aminoacyl-tRNA synthetase as a regulator of angiogenesis. Proc Natl Acad Sci U S A. 2002 Jan 8;99(1):173-7. Epub 2002 Jan 2.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/11773626>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC117534/>
 - Yang XL, Schimmel P, Ewalt KL. Relationship of two human tRNA synthetases used in cell signaling. Trends Biochem Sci. 2004 May;29(5):250-6. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/15130561>
-

Reprinted from Genetics Home Reference:

<https://ghr.nlm.nih.gov/gene/YARS>

Reviewed: January 2010

Published: March 21, 2017

Lister Hill National Center for Biomedical Communications

U.S. National Library of Medicine

National Institutes of Health

Department of Health & Human Services